



MPLKIP gene

M-phase specific PLK1 interacting protein

Normal Function

The *MPLKIP* gene (formerly known as *C7orf11*) provides instructions for making a protein called M-phase specific PLK1 interacting protein. The function of this protein is unclear. Based on its interaction with a protein called Plk1, the MPLKIP protein is thought to play a role in cell growth and division. In particular, it may help regulate the cell cycle, which is the cell's way of replicating itself in an organized, step-by-step fashion. Researchers speculate that the MPLKIP protein may also be involved in gene transcription, which is the first step in protein production.

Health Conditions Related to Genetic Changes

trichothiodystrophy

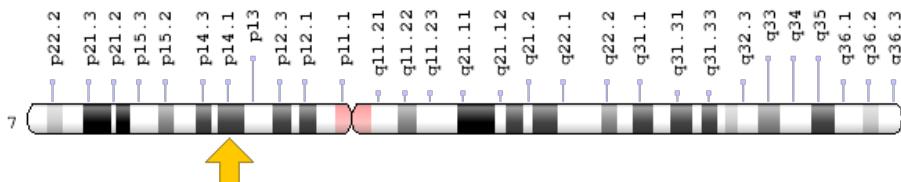
At least eight mutations in the *MPLKIP* gene have been identified in people with trichothiodystrophy. These mutations cause some cases of the non-photosensitive form of the disorder, which is not associated with extreme sensitivity to ultraviolet (UV) rays from sunlight.

All of the known *MPLKIP* gene mutations prevent the production of any functional MPLKIP protein. It is unknown how a loss of this protein leads to the characteristic features of trichothiodystrophy, including slow growth, intellectual disability, and brittle hair.

Chromosomal Location

Cytogenetic Location: 7p14.1, which is the short (p) arm of chromosome 7 at position 14.1

Molecular Location: base pairs 40,132,743 to 40,134,652 on chromosome 7 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ABHS
- C7orf11
- chromosome 7 open reading frame 11
- ORF20
- TTD non-photosensitive 1 protein
- TTDN1
- TTDN1_HUMAN

Additional Information & Resources

Educational Resources

- The Cell: A Molecular Approach (second edition, 2000): The Eukaryotic Cell Cycle
<https://www.ncbi.nlm.nih.gov/books/NBK9876/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28C7orf11%5BTIAB%5D%29+OR+%28MPLKIP%5BTIAB%5D%29%29+OR+%28TTDN1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- M-PHASE-SPECIFIC PLK1-INTERACTING PROTEIN
<http://omim.org/entry/609188>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_MPLKIP.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=MPLKIP%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=16002
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/136647>
- UniProt
<http://www.uniprot.org/uniprot/Q8TAP9>

Sources for This Summary

- Botta E, Offman J, Nardo T, Ricotti R, Zambruno G, Sansone D, Balestri P, Raams A, Kleijer WJ, Jaspers NG, Sarasin A, Lehmann AR, Stefanini M. Mutations in the C7orf11 (TTDN1) gene in six nonphotosensitive trichothiodystrophy patients: no obvious genotype-phenotype relationships. *Hum Mutat.* 2007 Jan;28(1):92-6.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/16977596>
- Faghri S, Tamura D, Kraemer KH, Digiovanna JJ. Trichothiodystrophy: a systematic review of 112 published cases characterises a wide spectrum of clinical manifestations. *J Med Genet.* 2008 Oct; 45(10):609-21. doi: 10.1136/jmg.2008.058743. Epub 2008 Jun 25. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/18603627>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3459585/>
- Hashimoto S, Egly JM. Trichothiodystrophy view from the molecular basis of DNA repair/transcription factor TFIIH. *Hum Mol Genet.* 2009 Oct 15;18(R2):R224-30. doi: 10.1093/hmg/ddp390. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19808800>
- Kraemer KH, Patronas NJ, Schiffmann R, Brooks BP, Tamura D, DiGiovanna JJ. Xeroderma pigmentosum, trichothiodystrophy and Cockayne syndrome: a complex genotype-phenotype relationship. *Neuroscience.* 2007 Apr 14;145(4):1388-96. Epub 2007 Feb 1. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17276014>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2288663/>

- Nakabayashi K, Amann D, Ren Y, Saarialho-Kere U, Avidan N, Gentles S, MacDonald JR, Puffenberger EG, Christiano AM, Martinez-Mir A, Salas-Alanis JC, Rizzo R, Vamos E, Raams A, Les C, Seboun E, Jaspers NG, Beckmann JS, Jackson CE, Scherer SW. Identification of C7orf11 (TTDN1) gene mutations and genetic heterogeneity in nonphotosensitive trichothiodystrophy. *Am J Hum Genet.* 2005 Mar;76(3):510-6. Epub 2005 Jan 11.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/15645389>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1196401/>
- Stefanini M, Botta E, Lanzafame M, Orioli D. Trichothiodystrophy: from basic mechanisms to clinical implications. *DNA Repair (Amst).* 2010 Jan 2;9(1):2-10. doi: 10.1016/j.dnarep.2009.10.005. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19931493>
- Zhang Y, Tian Y, Chen Q, Chen D, Zhai Z, Shu HB. TTDN1 is a Plk1-interacting protein involved in maintenance of cell cycle integrity. *Cell Mol Life Sci.* 2007 Mar;64(5):632-40.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17310276>

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/gene/MPLKIP>

Reviewed: May 2010

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services